CENTOGENE GUIDING PRECISION MEDICINE

CENTOGENE GmbH • Am Strande 7 • 18055 Rostock • Germany

Dr.
Institution
Address

Country

Order no.:
Order received: DD-MM-YYYY
Sample type / Sample collection date:
DNA / DD-MM-YYYY
Report date: DD-MM-YYYY
Report type: Final Report

Patient no.:, First Name:, Last Name:
DOB: **DD-MM-YYYY**, Sex:, Your ref.:

Test(s) requested: Epilepsy panel Upgraded

CLINICAL INFORMATION

Action tremor; Cerebellar atrophy; Dysarthria; Focal sensory seizure; Gait ataxia; Generalized-onset seizure; Headache
(Clinical information indicated above follows HPO nomenclature.)

Family history: Unknown.
Consanguineous parents: No.

POSITIVE RESULT
Pathogenic variant identified

INTERPRETATION

A heterozygous pathogenic variant was identified in the *CACNA1A* gene. **This result confirms the genetic diagnosis of autosomal dominant *CACNA1A*-related disorder.** Heterozygotes have a risk of 50% to transmit the variant to each offspring.

No further clinically relevant variants were detected.

RECOMMENDATIONS

- If possible, parental targeted testing is recommended as establishing the origin of the variant, inherited or *de novo*, is important for familial genetic counselling. Additionally, targeted testing for all affected family members, if any, is recommended.
- Genetic counselling is recommended.

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MAIN FINDINGS

SEQUENCE VARIANTS							
GENE	VARIANT COORDINATES	AMINO ACID CHANGE	SNP IDENTIFIER	ZYGOSITY	IN SILICO PARAMETERS*	ALLELE FREQUENCIES**	TYPE AND CLASSIFICATION***
CACNA1A	NM_023035.2:c.1748G>A	p.(Arg583Gln)	rs121908217	heterozygous	PolyPhen: Probably damaging SIFT: - MutationTaster: Disease causing Conservation_nt: high	gnomAD: 0.0000040 ESP: - 1000 G: -	Missense Pathogenic

Variant annotation based on CentoCloud Bioinformatics pipeline. * Splicing predictions: Ada and RF scores. ** Genome Aggregation Database (gnomAD), Exome Sequencing Project (ESP), 1000Genome project (1000G). *** Based on ACMG recommendations.

VARIANT INTERPRETATION

CACNA1A, c.1748G>A p.(Arg583Gln)

The *CACNA1A* variant c.1748G>A p.(Arg583Gln) causes an amino acid change from Arg to Gln at position 583 in exon(s) no. 13 (of 48). According to HGMD Professional 2025.4, this variant has previously been described as disease causing for Hemiplegic migraine and ataxia (PMID:10408534, 12707077, 23407676). ClinVar lists this variant (Interpretation: Pathogenic; Variation ID: 8505). It is classified as pathogenic based on CENTOGENE's implementation of the ACMG/AMP/ClinGen SVI guidelines.

Pathogenic variants in the *CACNA1A* gene are associated with different phenotypes. Developmental and epileptic encephalopathy-42 (DEE42) is a neurologic disorder characterized by the onset of various types of seizures in the first hours or days of life, although rare patients may have onset in the first weeks of life. The seizures tend to be refractory and associated with EEG abnormalities, including multifocal spikes and generalized spike-wave complexes. Affected infants show global developmental delay with severely impaired intellectual development. Other features may include axial hypotonia, peripheral hypertonia with hyperreflexia, tremor, ataxia, and abnormal eye movements (summary by the Epi4K Consortium, 2016; PMID:27476654). Mode of Inheritance: Autosomal dominant (OMIM®: 617106).

Episodic ataxia is a genetically heterogeneous neurologic condition characterized by spells of incoordination and imbalance, often associated with progressive ataxia. Mode of Inheritance: Autosomal dominant (OMIM®: 108500). Additionally, pathogenic variants in the *CACNA1A* gene are associated with spinocerebellar ataxia-6 (SCA6). Mode of Inheritance: Autosomal dominant (OMIM®: 183086).

Familial hemiplegic migraine-1 (FHM1) is an autosomal dominant form of migraine with aura. Typical attacks include a unilateral motor deficit associated with paresthesias, speech disturbances, or visual signs. These aura symptoms last from 10 minutes to a few hours and are followed by a migrainous headache. In some families, affected individuals have permanent cerebellar symptoms, such as nystagmus and slowly progressive mild to moderate statokinetic ataxia. In some cases, cerebral magnetic resonance imaging (MRI) reveals cerebellar atrophy (PMID: 9915947). Mode of Inheritance: Autosomal dominant (OMIM®: 141500).

METHODS

Epilepsy panel Upgraded
Genomic DNA is enzymatically fragmented, and target regions are enriched using DNA capture probes. These regions include approximately 41 Mb of the human coding exome (targeting > 98% of the coding RefSeq from the human genome build GRCh37/hg19), as well as the mitochondrial genome. The generated library is sequenced on an Illumina platform to obtain at least 20x coverage depth for > 98% of the targeted bases. CENTOGENE's in-house pipeline is applied. The sequencing reads are aligned to the Genome Reference Consortium Human Build 37 (GRCh37/hg19), as well as the revised Cambridge Reference Sequence (rCRS) of the Human Mitochondrial DNA (NC_012920). Sequence variants (SNVs/indels) and copy number variations (CNVs) are called using in house algorithms. Only the requested genes are analyzed; the panel gene list can be obtained in the appendix of this report. Variants with a minor allele frequency (MAF) of less than 1% in gnomAD database, or disease-causing variants reported in HGMD®, in ClinVar or in CENTOGENE's in-house Biodatabank are evaluated. The investigation for relevant variants is focused on coding exons and flanking +/-10 intronic nucleotides of genes with clear gene-phenotype

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evidence (based on OMIM® information). The value presented in the analysis statistics section is the sequencing coverage for coding exons and flanking +/-10 intronic nucleotides of the panel genes. All potential modes of inheritance are considered. In addition, the provided clinical information and family history are used to evaluate identified variants with respect to their pathogenicity and disease causality. Sequence variants are categorized into five classes (pathogenic, likely pathogenic, variant of uncertain significance [VUS], likely benign, and benign) according to ACMG/AMP guidelines for classification of variants in addition to ClinGen recommendations. CNVs are categorized into five classes (pathogenic, likely pathogenic, variant of uncertain significance [VUS], likely benign, and benign) according to ACMG/ClinGen guidelines for classification of CNVs. Additionally, other types of clinically relevant variants can be identified (e.g. risk factors, modifiers). All relevant variants related to the phenotype of the patient are reported. Likely benign and benign variants are not reported. CNVs of unknown significance with no apparent relation to the patient's phenotype are not reported. If applicable, mitochondrial variants with a heteroplasmy level of 15% or higher are reported. VUSs are usually not reported in the following cases: the described phenotype is already explained by detected pathogenic or likely pathogenic variants, the detected VUSs are not considered to be related to the described phenotype, there is a lack of clinical information, and/or the individual is asymptomatic or unaffected. CNV detection software has a sensitivity of more than 95%. CENTOGENE has established stringent quality criteria and validation processes for variants detected by NGS. Variants with low sequencing quality and/or unclear zygosity are confirmed by orthogonal methods. Consequently, a specificity of > 99.9% for all reported variants is warranted. Any detected CNV is only confirmed by an orthogonal method and reported when considered relevant to the phenotype.

ANALYSIS STATISTICS

Epilepsy panel Upgraded

Targeted nucleotides covered	≥ 20x	99.84%

LIMITATIONS

The genetic results are interpreted in the context of the provided clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the provided patient information or genetic data is inaccurate or incomplete. If the obtained genetic results are not compatible with the clinical findings, additional testing should be considered.

Epilepsy panel Upgraded
Only variants in genes potentially related to the proband's provided clinical information are reported. The genes with mapping issues in GRCh37/hg19 genome assembly, the non-protein-coding disease-associated genes, and approximately 0.2 Mb of genomic regions that are hard to sequence by current enrichment technology and are without evidenced relevance for monogenic disorders, are excluded from this analysis. More complex genetic events such as inversions, translocations, and repeat expansions, are not analyzed in this test. In addition, due to technology limitations, certain regions may be poorly covered, or not covered at all. In these regions and others encompassing repetitive, high-homology (such as pseudogene homology), and GC-rich sequences, relevant variants can be missed. Extremely low-coverage calls (homo/hemizygous or heterozygous calls with less than three or four reads, respectively) are expected to be artifacts based on our extensive validations and are consequently not considered during the analysis. Potential aberrant splicing is assessed with splice prediction tools. Intronic variants that are beyond 10 nucleotides from exon-intron boundaries are not considered for aberrant splicing analysis, with the exception of known pathogenic splicing variants evidenced by external sources. The CNV detection sensitivity is decreased for repetitive regions, homologous regions such as pseudogenes, and for events spanning 2 or less exons. Mitochondrial variants with heteroplasmy levels below 15% may not be detected. It is expected that lower quality samples (e.g., prenatal, product of conception, blood from patients with hematologic disorders, and highly degraded DNA) may generate lower quality NGS data; in these cases, CNV analysis and/or mitochondrial genome analysis may not be possible to perform.

ADDITIONAL INFORMATION

This test was developed and validated by CENTOGENE. It is intended for clinical purposes. All test results are reviewed, interpreted, and reported by our scientific and medical experts.

To exclude mistaken identity in sample collection, several guidelines recommend testing a second sample independently obtained from the consultand.

As variant classification can change over time, please feel free to contact CENTOGENE in the future to inquire about classification changes of reported variants.

DISCLAIMER

The preparation and processing of patient samples provided to CENTOGENE by a physician, clinical institution, or laboratory (by a "Partner"), as well as the genetic and/or biochemical testing requested, are performed according to the highest and most current scientific and analytical standards. However, in rare cases, genetic or biochemical tests may yield incorrect results—for example, due to the quality of the sample provided by the Partner, or due to unforeseeable or unknown factors beyond CENTOGENE's control. In such cases, CENTOGENE shall not be

held responsible or liable for incomplete, potentially misleading, or incorrect results if the issue could not have been identified in advance.

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APPENDIX

Epilepsy panel Upgraded

AARS1, AARS2, AASS, ABAT, ABCA2, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADGRG1, ADGRV1, ADPRS, ADSL, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGO1, AGPS, AIFM1, AIMP1, AIMP2, AKT1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANO4, ANTXR2, AP1G1, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APP, APTX, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARSA, ARSB, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP5F1A, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BAP1, BCAP31, BCKDHA, BCKDHB, BCORL1, BCS1L, BEST1, BET1, BLOC1S1, BLTP1, BOLA3, BORCS8, BRAF, BRAT1, BSCL2, BTD, C12orf57, C19orf12, C2orf69, CA5A, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2D, CAMK2G, CAMSAP1, CAPRIN1, CARS2, CASK, CASR, CAV1, CBL, CBS, CC2D2A, CCDC115, CCDC186, CCDC88A, CCDC88C, CCND2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHAMP1, CHD1, CHD2, CHD4, CHD5, CHKA, CHMP2B, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL11A2, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CREBBP, CRELD1, CRPPA, CSF1R, CSNK2A1, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHX16, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL1, DLL3, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, EMX2, ENTPD1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, F2, F5, FA2H, FAH, FAM50A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FBXO11, FBXO28, FDX2, FGF12, FGF13, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FTL, FUCA1, FUT8, FXYD2, FZR1, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GBE1, GCDH, GCH1, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GNAO1, GNAQ, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUSB, H3-3A, H3-3B, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ISCA2, ITPA, IVD, JAG1, JAM3, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KIFBP, KLHL20, KMT2E, KMT5B, KPTN, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LAT, LDB3, LETM1, LGI1, LIAS, LIPA, LIPT1, LIPT2, LMBRD2, LMNB1, LMNB2, LNPK, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MAST3, MBD5, MBOAT7, MCCC1, MCCC2, MCM3AP, MCOLN1, MDH2, MECP2, MECR, MED11, MED12, MED17, MED27, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPS22, MSX1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTOR, MTR, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYO5A, MYOT, NACC1, NAGA, NAGLU, NAGS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NCDN, NDE1, NDP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFE2L2, NFU1, NGLY1, NHLRC1, NOTCH1, NOTCH3, NPC1, NPC2, NPR2, NPRL2, NPRL3, NR4A2, NRAS, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OAT, OCLN, OCRL, OGDHL, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PGM2L1, PHACTR1, PHGDH, PHYH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLCG2, PLEKHG2, PLP1, PLPBP, PLXNA1, PMM2, PMP22, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPFIBP1, PPIL1, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRF1, PRICKLE1, PRICKLE2, PRMT7, PRODH, PRPF8, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSMB8, PSPH, PTCD3, PTCH1, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAC3, RAI1, RALA, RALGAPA1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNF216, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMD12, SAMHD1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SEC24D, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGCE, SGSH, SHH, SHQ1, SIK1, SIX3, SLC12A3, SLC12A5, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3, SLC39A8, SLC45A1, SLC4A10, SLC5A6, SLC6A1, SLC6A19, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMPD1, SMS, SNAP25, SNF8, SNX27, SON, SOX10, SPART, SPG11, SPG7, SPR, SPTAN1, SPTBN1, SPTBN4, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STAT1, STIL, STRADA, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEFM, TELO2, TET3, TFE3, TGFB1, TGIF1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM199, TMEM222, TMEM63B, TMEM70, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAF7, TRAK1, TRAPPC12, TRAPPC4,

TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRPS1, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTC19, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TYMP, TYROBP, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPB1, UQCRQ, USP18, USP7, VAMP2, VARS1, VARS2, VCP, VLDLR, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WFS1, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMIZ1, ZMYM2, ZNF142, ZNF335, ZNFX1

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